


Genome-wide association study

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 An abbreviated version of this protocol was published in Science Advances in Mar 2021

Nuclear genome-wide associations with mitochondrial heteroplasmy

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Detailed protocol

For mtDNA variants calling, referring to the supplementary methods:

"DNA extraction and genotyping were performed on saliva samples by National Genetics Institute (NGI), a CLIA licensed clinical laboratory and a subsidiary of Laboratory Corporation of America. The platform was a fully customized array with additional coverage of lower-frequency coding variation, and about 570,000 SNPs. Samples that failed to reach 98.5% call rate were re-analyzed. Individuals whose analyses failed repeatedly were re-contacted by 23andMe customer service to provide additional samples."

We used a custom Illumina custom Array (v4). Genotype calls are made using Illumina's Genome Studio software.

23andMe genotyping platforms	v1 (i.e. version 1): Illumina HumanHap550+ BeadChip
	v2: Illumina HumanHap550+ BeadChip
	v3: Illumina OmniExpress+ BeadChip
	v4: Illumina custom array
	v5: Illumina Infinium Global Screening Array

For mtDNA copy number estimates, papers from the literature suggest using median log-R ratio (Longchamps et al. 2020 <https://doi.org/10.1371/journal.pone.0228166>; Castellani et al. 2020 <https://doi.org/10.1186/s13073-020-00778-7>). Instead of median, we used mean, which was easier to track for the large amounts of data.

How to cite: (Readers should cite both the Bio-protocol preprint and the original research article where this protocol was used)

1. Nandakumar, P. , Hinds, D. and Sondheimer, N. (2021). Genome-wide association study. Bio-protocol Preprint. bio-protocol.org/prep1396.
2. Nandakumar, P., Tian, C., O'Connell, J., Hinds, D., Paterson, A. D. and Sondheimer, N.(2021). Nuclear genome-wide associations with mitochondrial heteroplasmy . Science Advances 7(12). DOI: [10.1126/sciadv.abe7520](https://doi.org/10.1126/sciadv.abe7520)

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